Claims

1. A method for the diagnosis of SMEI in a patient comprising:

(1) detecting an alteration in the SCN1A gene, including in a regulatory region of the gene, in a patient sample;

(2) ascertaining whether the alteration is known to be SMEI associated or non-SMEI associated; and

(3) (a) establishing a diagnosis of a high probability of SMEI where the alteration is known to be SMEI associated; or

(b) establishing a diagnosis of a low probability of SMEI where the alteration is non-SMEI associated; or

(d) or, if not known to be either,

(i) considering genetic data for parents and/or relatives;

(ii) establishing whether the alteration has arisen *de novo* or is inherited; and

(iii) establishing a diagnosis of a low probability of SMEI where the alteration is inherited but a diagnosis of a high probability of SMEI if the alteration is *de novo*.

2. A method as claimed in claim 1 further comprising establishing whether the alteration would result in a major disruption to the protein and, if *de novo*, establishing a diagnosis of a very high probability of SMEI.

3. A method as claimed in claim 2 wherein the alteration is a truncating mutation.

4. A method as claimed in claim 1 wherein the alteration is one identified in Table 3 as SMEI associated or non-SMEI associated.

5. A method as claimed in claim 1 comprising performing one or more assays to test for the existence of an SCN1A alteration and to identify the nature of the alteration.

6. A method as claimed in claim 5 comprising:

(1) performing one or more assays to test for the existence of an alteration in the SCN1A gene of the patient; and if the results indicate the existence of an alteration in the SCN1A gene;

(2) performing one or more assays to identify the nature of the SCN1A alteration.

7. A method as claimed in claim 5 wherein one of the assays is a DNA hybridisation assay.

8. A method as claimed in claim 7 wherein an SCN1A gene probe, an SCN1A exon-specific probe, or an SCN1A allele specific probe is hybridised to genomic DNA isolated from said patient.

9. A method as claimed in claim 5 wherein one of the assays is high performance liquid chromatography.

10. A method as claimed in claim 5 wherein one of the assays is an electrophoretic assay.

11. A method as claimed in claim 5 wherein the sample DNA to be tested is quantitatively amplified for at least one exon of the SCN1A gene to produce amplified fragments and the length of the amplification products for each amplified exon is compared to the length of the amplification products obtained when a wild-type SCN1A gene is amplified using the same primers, whereby differences in length between an amplified sample exon

and the corresponding amplified wild-type exon reflect the occurrence of a truncating alteration in the sample SCN1A gene.

12. A method as claimed in claim 5 wherein one of the assays incorporates DNA amplification using SCN1A allele specific oligonucleotides.

13. A method as claimed in claim 5 wherein one of the assays is SSCP analysis.

14. A method as claimed in claim 5 wherein one of the assays is RNase protection.

15. A method as claimed in claim 5 wherein one of the assays is DGGE.

16. A method as claimed in claim 5 wherein one of the assays is an enzymatic assay.

17. A method as claimed in claim 16 wherein said assay incorporates the use of MutS.

18. A method as claimed in claim 5 wherein one of the assays examines the electrophoretic mobility of the SCN1A protein of the patient.

19. A method as claimed in claim 5 wherein one of the assays is an immunoassay.

20. A method as claimed in claim 5 wherein one of the assays is DNA sequencing.

21. A method for the diagnosis of SMEI in a patient, comprising:

detecting an alteration in the SCN1A gene, including in a regulatory region of the gene, in a

patient sample, and establishing a diagnosis of a high probability of SMEI if a SMEI associated alteration as laid out in Table 3 is identified or, in the alternative, establishing an a diagnosis of a low probability of SMEI if a non-SMEI associated alteration as laid out in Table 3 is identified.

22. A method of determining the appropriate treatment for a SMEI patient comprising performing the method claimed in claim 1 or 21 and correlating the diagnosis reached with known indications and contra-indications for SMEI patients.

23. A method of determining the likelihood of adverse results from treatments of a SMEI patient including drug treatments and vaccinations comprising performing the method as claimed in claim 1 or 21 and correlating the diagnosis reached with known indications and contra-indications for SMEI patients.